GALACTOSEMIA

The most common form of galactosemia, a metabolic disorder, is the result of very little or absence of the enzyme galactose-1-phosphate uridyl transferase. This enzyme is involved in the digestion of galactose, a breakdown product of lactose, which is present in milk, most infant formulas, and naturally occurring in organ meats, legumes, fruits, and vegetables. Deficiency of galactose-1-phosphate uridyl transferase quickly raises the galactose content in the blood to dangerous levels. Screening in Connecticut was instituted in 1964.

GENETIC BASIS

- Autosomal recessive condition—Recurrence risk is 25%
- Prenatal diagnosis is possible

CT PREVALENCE 1:47,546

CLINICAL FEATURES OF UNTREATED DISEASE

Severity

- Physical disabilities: cerebral palsy, ataxia, seizures, cataracts, liver disease
- Developmental disabilities: mental retardation
- Mortality: liver failure, sepsis, or bleeding can cause severe morbidity and death

Symptomatic diagnosis

Symptoms usually occur within the first two weeks of life and may even occur before receiving the results of newborn screening. Early symptoms include jaundice, vomiting, lethargy, hepatosplenomegaly, cataracts, failure to thrive, hypoglycemia, and sepsis.

Variants

Duarte Galactosemia is a milder form where activity of galactose-1-phosphate uridyl transferase is about 25-50% of the enzyme’s normal activity. Research has not revealed medical or other developmental complications associated with Duarte Galactosemia.

CLINICAL OUTCOME WITH SCREENING AND TREATMENT

Mortality

Reduced substantially, but some infants may die before the results of the screening test are available because of susceptibility to E. coli septicemia.

Clinical Disability

IQ levels may be effected. The best results (normal IQ) have been obtained when treatment is started before 10 days of age. Visual perception, speech, and other learning disabilities are common even in early treated infants. Ovarian failure with hypergonadotropic hypogonadism and primary or second amenorrhea has occurred in the majority of females.

Variability

Phenotypic and genetic classification is necessary to determine treatment and compare outcomes.
Interventions
A galactose-restricted diet should begin as soon as possible and should continue throughout life.
A nutritionist and metabolic specialists must coordinate therapy.

SCREENING FOR GALACTOSEMIA

Laboratory tests
- Blood spots are tested for galactose-1-phosphate uridyl transferase utilizing “Neonatal Chemistry System” (NCS) – Quantitative Fluorometric Testing. If there is no activity, a test for total galactose is performed.

Abnormal test results
- The primary care provider is notified by the DPH, Genetic NBS Tracking Unit nurse consultants.
- A referral is made by the DPH, Genetic NBS Tracking Unit nurse consultant to one of the Genetic Regional Treatment Centers.
- The primary care provider will be advised by the DPH, Genetic NBS Tracking Unit nurse consultant to contact the Genetic Regional Treatment Center to make a prompt referral and arrange for confirmation testing and evaluation.

The information provided is offered for general informational and educational purposes only. It is not offered as and does not constitute medical advice. In no way are any of the materials presented meant to be a substitute for professional medical care or attention by a qualified practitioner, nor should they be construed as such. Contact your physician if there are any concerns or questions.

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